

[Click here to view linked References](#)**Supplementary Table 1 Clinical characteristics of patients in group I to VI**

Patient no	Gender	Age at diagnosis	MSI	IHC loss	Am criteria	No of polyps	CRC	Location	Stage	Other
<b>I) CRC familial or unknown inheritance not polyposis</b>										
1	f	75	MSS	-	yes	-	yes	sigmoid	-	
2	f	26	MSS	-	no	-	yes	cecum	II	
3	m	60	MSS	-	no	-	yes	sigmoid	III	
4	m	27	MSS	-	yes	-	yes	-	II	ulcerative colitis at 10 years
5	f	41	MSS	-	yes	-	yes	sigmoid	II	
6	f	41	MSS	-	no	-	yes	-	II	breast cancer
7	f	35	MSS	-	no		yes	rectum	III	
8	f	37	MSS	-	no	-	yes	rectum	III	
9	f	62	MSS	-	no	-	yes	appendix	-	
10	f	58	MSS	-	yes	-	yes	rectum	-	
11	f	42	MSS	-	yes	-	yes	rectum	-	
12	f	-	MSS	-	yes	-	yes	rectum	-	
13	f	33	MSI low	-	no	-	yes	-	-	
14	f	51	MSS	-	no	-	-	sigmoid	-	breast cancer
15	f	52	MSS	-	no	1-100	-	-	-	endometrial cancer
16	f	82	MSS	-	yes	-	-	cecum	-	
17	f	46	MSS	-	yes	-	yes	rectum	III	
18	f	53	MSS	-	no	1-100	yes	-	III	
19	f	40	MSS	-	yes	1-100	yes	-	III	
20	m	76	MSS	-	no	1-100	yes	sigmoid	-	
21	f	38	MSS	-	no	1-100	yes	sigmoid	II	
22	m	74	MSS	-	yes	-	yes	rectum	-	
23	m	48	MSS	-	yes	-	yes	cecum	II	

24	m	60	MSS	-	yes	1-100	yes	sigmoid	-	
25	f	51	MSS	-	yes	1-100	yes	-	I	
26	f	50	MSI-H	-	no	-	yes	-	-	endometrial cancer
27	f	40	MSI-H	-	no	1-100	yes	ascending	II	sigmoid polyps
28	m	41	MSS	-	no	-	yes	transverse	-	
29	m	31	MSS	-	no	-	-	-	-	
30	f	32	MSS	-	no	-	yes	-	-	
31	f	23	MSS	-	no	-	yes	sigmoid	III	
32	f	31	MSS	-	yes	-	yes	-	-	
33	f	51	MSS	-	-	-	yes	-	-	
34	f	66	MSI-H	MLH1/PMS2	no	-	yes	caecum	II	
35	f	61	MSI-H	MLH1/PMS2	no	-	yes	caecum	II	
36	m	51	MSI-H	MLH1/PMS2	no	-	yes	rectum	II	rectal cancer
37	f	70	MSI-H	MLH1/PMS2	no	-	yes	transverse	II	endometrial cancer
38	f	63	MSI-H	MLH1/PMS2	no	-	no	-	I	endometrial cancer
39	m	49	MSI-H	MLH1/PMS2	no	-	yes	ascendens	II	
40	f	61	MSI-H	-	no	-	yes	ascendens	II	
41	f	57	MSI-H	MLH1/PMS2	yes	-	yes	ascendens	III	
42	f	50	MSI-H	MLH1/PMS2	no	-	yes	ascendens	II	
43	f	41	MSI-H	MLH1/PMS2	no	-	yes	transverse	II	
44	f	38	MSI-H	MLH1/PMS2	no	-	yes	ascendens	IV	breast cancer, 32 years
45	f	40	MSI-H	MLH1/PMS2	yes	-	no	-	I	endometrial cancer
46	f	57	MSI-H	MLH1/PMS2	no	-	no	-	I	endometrial cancer
47	f	68	MSI-H	MLH1/PMS2	no	-	yes	ascendens	II	endometrial cancer, 53 years
48	f	55	MSI-H	MSH6	no	-	yes	ascendens	II	synchronous endometrial cancer, 55 years
49	f	21	MSI-H	MSH6	no	-	yes	rectum	III	childhood brain tumour
50	f	80	MSI-H	MSH6	yes	-	yes	ascendens	II	endometrial cancer, 50 years

51	f	35	MSI-H	-	no	-	yes	rectum	III	
52	m	76	MSS	-	yes	-	yes	ascendens	II	
54	f	48	MSI-H	-	no	-	-	-	I	endometrial cancer
55	f	79	MSS	-	no	-	yes	rectum	II	rectal cancer
56	m	54	MSI-H	-	yes	-	yes	rectum	I	rectal cancer
57	m	41	MSI-H	-	no	-	yes	ascendens	III	
91	m	39	MSI-H	-	yes	-	yes	-	-	
92	f	45	MSI-H	-	no	-	yes	-	-	endometrial cancer, 45 years, breast cancer, 53 years

#### II) Unexplained adenomatous polyposis >100 polyps, inheritance

58	f	17	-	-	-	>1000	no	-	-	
59	f	31	-	-	-	101-1000	yes	descending	-	several family members with polyps and crc

#### III) Unexplained adenomatous polyposis 1-100 polyps, inheritance

60	-	21	-	-	-	1-100	-	-	-	
61	m	-	-	-	-	-	-	-	-	
62	f	38	-	-	-	1-100	-	descending	-	
63	f	45	MSS	-	-	1-100	-	sigmoid	-	
64	m	35	-	-	-	1-100	-	-	-	tubular polyps
65	m	66	-	-	-	1-100?	yes	-	-	
66	f	41	-	-	-	1-100	yes	rectum	-	sister of the paternal grandmother with breast cancer, 31 years
67	f	41	-	-	-	1-100	yes	-	-	

#### IV) Unexplained adenomatous polyposis unknown inheritance

68	f	84	-	-	-	1-100	-	-	-	
69	m	48	-	-	-	1-100	yes	rectum	-	duodenal polyp
70	m	57	-	-	-	1-100	yes	descending	-	FGP
71	f	47	-	-	-	1-100	yes	sigmoid (x2)	-	flat adenomas
72	m	31	-	-	-	1-100	no	-	-	FGP

73	m	58	-	-	-	1-100	no	-	-	
74	f	46	-	-	-	1-100	yes	-	-	
75	m	60	-	-	-	1-100	yes	sigmoid	-	
76	f	52	-	-	-	1-100	yes	descending	-	

**V) Familial or simplex atypical polyposis/mixed polyposis/serrated polyposis**

77	f	60	-	-	-	1-100	yes	rectum	-	atypical polyposis
78	f	50	-	-	-	1-1000	yes	rectum	-	
79	f	50	-	-	-	1-100	yes	descending (x3)	-	
80	f	51	-	-	-	1-100	no	-	-	duodenal polyps
81	f	11	-	-	-	1-100	-	-	-	no inheritance
82	m	45	MSS	-	no	1-100	-	-	-	
83	f	54	-	-	-	1-100	yes	-	-	breast cancer, 59 years
84	f	36	-	-	-	100-200	-	-	-	mixed polyposis
85	m	71	-	-	-	1-100	-	-	-	several family members and both parents with crc
86	m	74	-	-	-	1-100	-	-	-	
87	m	<50	-	-	-	1-100	-	-	-	mixed polyposis
88	f	-	-	-	-	1-100	-	-	-	mixed polyposis

**VI) Polymerase proofreading associated polyposis (PPAP)**

89	f	-	MSS	-	yes	1-100	yes	-	-	See Rohlin et al 2014
90	m	43	MSS	-	no	-	yes	sigmoid	III	See Rohlin et al 2016

neg-negative, Am-Amsterdam, FGP-fundic gland polyps, IHC-immunohistochemistry, MSS-microsatellite stable, MSI-microsatellite unstable